Specialty Conference

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Spinal Dysraphism

An Interdisciplinary Diagnostic Approach

WILLIAM L. NYHAN, MD, PHD:* *I am pleased to introduce Dr. Hector James, who is Assistant Professor of Neurosurgery and Pediatrics. He joined the faculty at the University of California, San Diego, this year and is the director of Pediatric Neurosurgery. He is going to speak on spinal dysraphism.*

HECTOR E. JAMES, MD:† I would like to cover this topic by defining the clinical entities, the clinical presentation of the syndromes, a comprehensive diagnostic approach using new modalities, and finally therapy.

Under the term spinal dysraphism popularized by Lichtenstein[1] one can place a large family of disorders. However, common usage, primarily in Great Britain, has been to reserve the term for pathologic conditions of the spine that are not overt, excluding myelomeningocele, for instance, or spina bifida cystica. The term implies a problem that presents primarily as an occult syndrome, a disorder that is not evident at birth unless specifically sought. The pathogenesis and the clinical syndromes have been well defined by James and Lassman.[2]

Pathogenesis may occur in two different forms. One is the *pressure group*. If, for instance, a lipoma is growing inside the spinal canal there will be a dysfunction of the conus medullaris or nerve roots because of compression in the rigid canal of the neural elements, as the mass continues growing inside it. The other is the *traction group*. Normally the spinal cord migrates with age because of differential rates of growth between the spinal column and the spinal cord itself. If this migration is interfered with, traction of the neural elements of the conus medullaris and spinal cord can occur. From Barson's[3] analysis of the position of the conus medullaris in the spinal canal of human fetuses at different stages of gestation, the normal adult position for the conus medullaris is not reached until after birth. There is a progressive migration of the conus medullaris during gestation because the spinal column is growing at a much faster rate than the spinal cord. The final position of the conus medullaris may not be reached until the fourth or fifth year of life. What types of pathologic conditions can interrupt this migration? In Tills'[4] series the vast majority are situations in which there is tethering of the conus. When this happens there is traction from the filum terminale to the conus medullaris and normal migration is impaired. Next most common is diastematomyelia. In this condition a spicule of bone or connective tissue without bone transects the spinal canal from its ventral to dorsal aspect, and in so doing transects the spinal cord and restricts rostral migration. In Matson's[5] series, the most common site is in the lumbar region; more rarely it occurs in the thoracic region.

Clinical Presentation

James and Lassman[2] defined two major clinical presentations, though combinations of both in

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TABLE 1.—*Spinal Dysraphism*

Clinical syndrome
Characteristically slow clinical course
Orthopedic syndrome
Cutaneous anomalies (may be present)
Asymmetry of legs
Diminished muscle bulk
Shortened foot
Inversion of foot, pes cavus, elevation of the first metatarsal head, clawing of toes
Peculiar gait
Reflex changes may be present, usually late
Urological syndrome
Bladder changes: alterations in urinary frequency, retension, incontinence
With or without bowel symptoms
With or without cutaneous lesions
Tethered cord syndrome
Terminology used to cover the above spectrum

which one or the other is dominant are more common (Table 1). The first is the orthopedic syndrome in which there are cutaneous anomalies such as exuberant growth of hair, telangiectasis, or a small lipoma underneath the skin—something that will signal the disorder below it. Asymmetry of the legs is characteristic. There is diminished muscle bulk and a shortened foot. When you look closely at the foot you may find the following: inversion of the foot, a pes cavus deformity, elevation of the metatarsal head, or clawing of the toes. These changes result from an imbalance between agonist and antagonist muscles.

The second is the urological syndrome. In a child with a previously normal voiding pattern and continence, a disorder subsequently develops in which there is frequency of urination or incontinence, or repeated urinary tract infections occur not readily explained by urologists. Designation of the syndromes in this way[2] is useful because it alerts us to the populations of patients that should be considered for this diagnosis. A child with the orthopedic syndrome is usually referred from the pediatrician or the family practice physician to the orthopedic surgeon, who lengthens or shortens tendons to make the limb look better. Finally, when the disorder progresses despite surgical procedures the child is referred to a neurosurgeon. The child with the urological syndrome is commonly referred to a urologist because of an abnormality in voiding pattern or urinary tract infections. After repeated normal findings on cystoscopies and radiologic studies, it finally becomes apparent that something is wrong with the innervation of the bladder and sphincter mechanism.

Unfortunately we tend in medical education to emphasize those things that are common and, often, obvious. Among the disorders of the spinal cord, therefore, we devote considerable attention to myelomeningocele and related conditions that are easy to diagnose. A general physician, orthopedist or urologist often has had no experience with the disorders we are discussing today, and he may not have been taught that they exist. Therefore the patient often reaches a neurosurgeon late in the disease. In reality these are the conditions we want to see the earliest, because it is at that time that therapy is of most value. We are dealing with central nervous system axons and neurons and damage once done is frequently irreversible. The value of corrective surgical therapy is that it prevents progression.[4,6] I will present two cases as examples of this.

CASE 1. The mother of a 5-year-old boy had noted when the child was 6 months old that there was an asymmetry of the lower extremities. As commonly happens, she was not listened to. We saw the child after two tendon procedures had been done. The form of presentation, therefore, was the orthopedic syndrome. The findings were wasting of the gastrocnemius on one side and gross asymmetry of the feet, one being smaller than the other. When we looked more closely at the smaller foot we could see a pes cavus deformity, elevation of the first metatarsal head and clawing of the toes. He had a diastematomyelia.

CASE 2. The patient was seen at birth by two physicians who insisted, contrary to the mother's view, that the child's footprints were normal. The boy finally was brought to a pediatrician who referred him to us. There was no doubt that the mother was correct. There was something wrong with the footprint. The footprint was that of a patient with the orthopedic syndrome. There was a cavus deformity with a lack of imprint in the area of the elevated head of the first metatarsal, and the toes came down as a claw. This child had a filum terminale tethered to a skin dimple in the coccygeal region.

One must differentiate these disorders that are more common from another group of disorders called spinal hamartomas.[7] These latter present very commonly with a lump in the back, and the differential diagnosis ranges from myelomeningo-

cele which is not possible because the skin is intact, to sacrococcygeal teratoma. The growths are called hamartomas because the tissue inside them is adult mature tissue; it is simply out of its proper place. Palpable bone may be felt in the mass. Results of neurological examination at birth may be normal. We have warned[7] that if the entity is not promptly treated traction can occur. Surgical therapy, therefore, is indicated for these hamartomatous conditions.

Diagnosis

Clinical findings, history and physical examination are as always imperative elements in coming to the diagnosis. The diagnostic studies that are done routinely include x-ray studies of the spine. One may see a variety of things in the spine, abnormal structures with punched out areas in the lamina where mesodermal fusion in the midline was inadequate, or a slightly widened spinal canal with a spicule of bone inside it. This is the classic appearance of diastematomyelia. If it is present, no further studies are needed. Unfortunately there is so much cartilage in this area and poorly calcified bone in a young child that the radiological report is often that of a normal spine, because the anomaly cannot be seen. Myelography has been the classical approach to the diagnosis of intraspinal problems but we are getting away from it. Pediatricians do not like to refer children for invasive procedures, and I agree with them. Then, to do a myelogram in a child, general anesthesia is often required. Also, most radiologists have had little experience with the performance of myelograms in children because this procedure is not often done. Finally, there may be a very low-set conus medullaris, and a lumbar puncture and injection of contrast agent may be a dangerous way to study the area. Alternatively, if one tries the cisternal route or the lateral cervical approach, there could be the problem of an Arnold-Chiari malformation and a descended brain stem, and again harm could occur.[8]

It became evident to us that the techniques employed in the diagnosis of these syndromes were not at all satisfactory. In the presence of an enormously distended bladder, there is no problem leading one to suspect a neurologic abnormality. It is in borderline situations that one needs help for early detection. There are usually no significant changes in findings on the intravenous pyelogram. Fortunately the urologist can be of assistance using urodynamic studies. These consist in the monitoring of the rectal sphincter tone and the rectal electromyogram (EMG), simultaneously with the filling of the bladder, the classical cystometrogram. The method employs a catheter with a double lumen; one lumen is to inject air to inflate the bladder and the other is to record pressure. At the same time, as the catheter is pulled out using timed technique, one can monitor the pressure of the internal and external sphincters. Since the dermatomes that innervate the rectal sphincter also innervate the sphincter of the bladder, one may see if the reflex arc is intact or if it is impaired. With the advent of this technique, we began to detect problems very early in their development.

The other factor that needed improvement was the diagnosis of changes inside the spinal canal, preferably using a noninvasive procedure. The advent of computer tomography (CT) has permitted this. CT can assess the type of tissue we are dealing with using what are called the Hounsfield numbers. These are the quantitative representation of the density of absorption of the radiation source. As computer technology improved, magnification of CT scans became feasible. We then looked at the myelographic criteria that were used to diagnose a tethered conus. Gryspeerdt[8] defined three primary criteria: a low position of the conus medullaris; a dorsally located filum terminale (normally there is lumbar lordosis and if the filum is tethered because of the curvature of the spine, it will be located in the dorsal aspect of the canal), and, finally, a filum terminale that has a width greater than 2 mm.

With these criteria we proceeded to study the CT data. We had an unusual opportunity with a two-year-old girl whose foster mother became quite concerned because the child started to walk late. Moreover, when the child did start to walk she had an obvious limp on the left. The same pediatrician that referred the previous child with the abnormal footprint saw her, found a sacral skin dimple, and referred her as a potential patient with a tethered cord. We noted a smaller foot and smaller muscle bulk of the gastrocnemius on the left side. Her bowel and bladder function seemed to be intact and passed urodynamic testing. Roentgenograms of the spine showed some widening of the lumbosacral junction and of the sacrum itself, though they had

been reported as normal. A CT of the spine was requested. In the study we could see the iliac crest, the sacrum, the bifid sacrum heralding a mesodermal disorder, and a structure inside the sacrum at S1 which should not have been there. We then set the scanner so that the computer would analyze the tissue that was in the sacrum, and it indicated that it was fat. We then analyzed the tissue inside the round element that was in the canal, and the computer read 16 Hounsfield units, which indicated fibrous or glial tissue

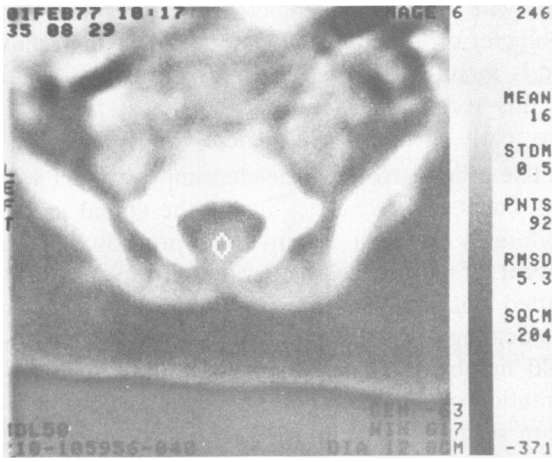

Figure 1.—Computer tomography scan through the upper sacrum. There is a rounded structure, dorsally located and forming a unit with the midline defect of the lamina. The Cursor reading of the scanner identifies fibrous-glial tissue (16 Hounsfield units, upper right corner). (Reproduced by permission from James HE, Oliff M: Computer tomography in spinal dysraphism. J Computer Assisted Tomography 1:391-397, 1977, Raven Press, New York.)

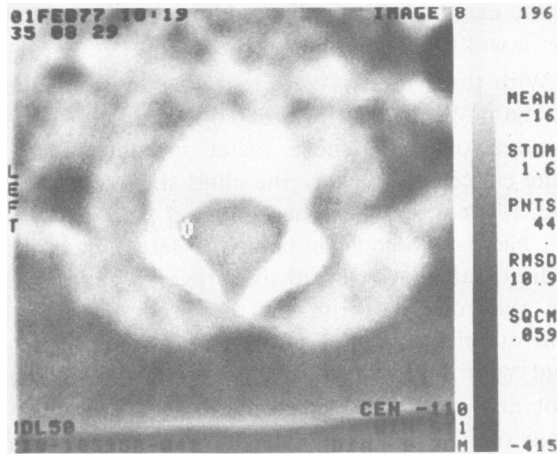

Figure 2.—Computer tomography scan of the same patient as in Figure 1, at the L4 level, showing a low set conus in the spinal canal, allowing little room for the normally extensive epidural fat (Cursor reading: −16 Hounsfield units, upper right corner).

(Figures 1 and 2). We then had a rounded structure that was centrally and slightly dorsally located in the sacrum that was continuous with the defect in the back of the sacrum. We therefore had the first two of Gryspeerdt's criteria.[8] There is nothing that should be rounded, plump and located dorsally in the canal except a thickened filum terminale. What we would like next is the third criterion, that is its transverse diameter. So we turned the computer to what is called the "F" series, which allows us to measure between two points on each side of the filum, and it read 8.93 mm. Anything measuring more than 2 mm is a thickened filum. At operation we confirmed these findings. We sectioned the filum terminale at its point of attachment.

Let us discuss two more examples.

CASE 3. An adolescent had an absent ankle jerk on the left, wasting of the left gastrocnemius, and a lump off the midline in the sacral area. On CT examination the normal paraspinal musculature, subcutaneous fat and a bifid vertebral body at the level of L5 were evident. The paraspinal muscles were no longer symmetrical, and there was a local increase in subcutaneous fat. The upper sacrum was bifid, and we could follow the subcutaneous fat into the area of the canal. This was a leptomyelipoma in a bifid spine, an occult presentation of a lipoma that was tethering the cord and invading the spinal canal.

CASE 4. The next case was a 9-month-old child of a diabetic mother. The child had a peculiar looking pelvis, as well as wasting of one foot and of the gastrocnemius on one side. It was felt that the child had a pathologic condition of the spinal cord. On roentgenographic examination the iliac crests were close together, indicating an agenesis of the sacrum. The CT scans started at L2 and proceeded down into the sacrum. We saw a remarkable narrowing of the spinal canal. These children can have lumbar stenosis and compression of the nerve roots and conus medullaris. As the study continued the normal vertebral bodies disappeared, and we could see the iliac crests on each side. Bone had been replaced by fat.

In summary, what has been attained over the past several years is an interdisciplinary approach to the recognition of the clinical entity and the application of new techniques in urology and radiology.[9] We now have a noninvasive diagnostic approach which facilitates early appropriate therapy for this disease.

DR. NYHAN: *Are there questions?*

QUESTION: *Would you carry out studies in every child with a sacral dimple?*

DR. JAMES: That is a very good question. In the article by Powell and associates[10] a large number of children with dimples were studied and only two or three had any neurological problem. The dimple becomes important when there is evidence of any of the clinical components of either the urological or the orthopedic syndromes. We do not do tests in every child with a dimple in the back, but what we do is listen to the mother's story and examine the child. If the question is then still present there is no reason why further studies should not be done.

QUESTION: *How about children with midline lumbar or sacral masses?*

DR. JAMES: If there is any sort of lump I would become extremely concerned. That is a clear heralding sign of an underlying mesodermal disorder. The ectoderm can have disorders on its own without significant underlying problems. The mesoderm seems to be the primary component in all of these disorders. Any child with a lump in this area needs a thorough evaluation.

One thing I would like to mention in closing is that until June 1977 we had placed these disease entities in a separate category from those of myelomeningoceles. There is now new information on the familial incidence of these diseases. At the June 1977 meeting of the International Society for Pediatric Neurosurgery, Till[11] stated that statistical analyses from London showed a higher incidence of dysraphic disorders, including myelomeningoceles, in families in which there have been occult forms of dysraphism. Alpha-fetoprotein determination on amniotic fluid is now recommended in subsequent pregnancies in these families.

REFERENCES

1. Lichtenstein BW: Spinal dysraphism. Arch Neurol Psychiatry 44:792-810, 1940

2. James CCM, Lassman LP: Spinal dysraphism. Arch Dis Child 35:315-327, 1960

3. Barson AJ: The vertebral level of termination of the spinal cord during normal and abnormal development. J Anat 106:489-497, 1970

4. Till K: Spinal dysraphism: A study of congenital malformations of the lower back. J Bone Joint Surg (Brit) 51:415-422, 1969

5. Matson DD: Spinal bifida occulta, *In* Neurosurgery of Infancy and Childhood, 2nd Ed. Springfield, IL, Charles C Thomas, 1969, pp 9-21

6. Thomas JE, Miller RH: Lipomatous tumors of the spinal canal: A study of their clinical range. Mayo Clin Proc 48:393-400, 1973

7. Tibbs PA, James HE, Rorke LB, et al: Midline hamartomas masquerading as meningomyeloceles or teratomas in the newborn infant. J Pediatr 89:928-933, 1976

8. Gryspeerdt GL: Myelographic assessment of occult forms of spinal dysraphism. Acta Radiol (Diagn) 1:702-717, 1963

9. James HE, Oliff M: Computed tomography in spinal dysraphism. J Computer Assisted Tomography 1:391-397, 1977

10. Powell KR, Cherry JD, Hougen TJ, et al: A prospective search for congenital dermal abnormalities for the craniospinal axis. J Pediatr 87:744-750, 1975

11. Till K: Spinal dysraphism. Meeting of the International Society for Pediatric Neurosurgery, June 22-25, 1977, Guaruja, Brazil